

CLAIMS

1. A pharmaceutical composition comprising a substance capable of causing an aberrant gene product to function in a manner similar to the wild-type gene product and a pharmaceutically acceptable carrier.
2. The pharmaceutical composition according to claim 1, wherein the substance is an agonist or an antagonist of the aberrant gene product.
3. The pharmaceutical composition according to claim 1, wherein the aberrant gene product is an aberrant receptor, an aberrant channel, an aberrant signal or an aberrant enzyme.
4. The pharmaceutical composition according to claim 1, wherein the aberrant gene product is an aberrant receptor and wherein the composition is used for prophylaxis and/or treatment of a disease caused by an aberrant receptor.
5. The pharmaceutical composition according to claim 4, wherein the disease caused by an aberrant receptor comprises a disease caused by substantial reduction in the activity of the signal transduction system of the cell having the aberrant receptor.
6. The pharmaceutical composition according to claim 4, wherein the disease caused by an aberrant receptor comprises a disease caused by the substantial absence of the action of a natural ligand on the signal transduction system of the cell having the aberrant receptor.
7. The pharmaceutical composition according to claim 4, wherein the disease caused by an aberrant receptor comprises a disease caused by substantial reduction in the affinity of a natural ligand for the aberrant receptor.
8. The pharmaceutical composition according to claim 6, wherein the signal transduction system comprises a signal transduction system based on the change in intracellular concentration of a responding substance resulting from the binding of a natural ligand and a receptor.

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13. The use according to claim 12, wherein the aberrant gene product is an aberrant receptor.

15. The screening method according to claim 14, wherein the aberrant gene product is an aberrant receptor.

17. A method of screening for a drug for restoring normal function to a signal transduction system of a cell having an aberrant receptor of a mammal suffering from a disease caused by the aberrant receptor, which comprises bringing the aberrant receptor into contact with a subject substance and assaying the activity of said substance on said

receptor and wherein the activity is an activity that restores the normal function of the cell.

18. The screening method according to claim 16, wherein the aberrant receptor is an aberrant receptor prepared by
5 expressing in a cell the gene encoding the aberrant receptor.

19. The screening method according to claim 16, wherein the gene encoding the aberrant receptor is an aberrant receptor-encoding gene specified by comparative analysis of
10 a gene prepared from a cell of a mammal suffering from a disease caused by the aberrant receptor, and a gene prepared from a cell of a mammal of the same species that does not carry the aberrant receptor.

20. A method of preparing a drug for treatment of a disease
15 caused by an aberrant gene product, which comprises bringing the aberrant gene product into contact with a subject substance, assaying the activity of said substance on said product and preparing a substance judged to substantially operate the signal transduction system of a
20 cell having the aberrant gene product wherein said activity is activity that restores wide-type activity of the gene product.

21. A method of preparing a substance for treatment of a disease caused by an aberrant receptor, which comprises
25 bringing the aberrant receptor into contact with a subject substance, assaying the activity of said substance on the aberrant receptor and preparing a substance judged to substantially operate the signal transduction system of a cell having the aberrant receptor, wherein said activity is
30 activity that restores wild-type activity to the receptor.

22. The method according to claim 21, wherein the aberrant receptor is an aberrant receptor prepared by expressing in a cell the gene encoding the aberrant receptor.

23. The method according to claim 22, wherein the gene
35 encoding the aberrant receptor is an aberrant receptor-encoding gene specified by comparative analysis of a gene

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prepared from a cell of a mammal suffering from a disease caused by the aberrant receptor, and a gene prepared from a cell of a mammal of the same species that does not carry the aberrant receptor.

5 24. A method of screening for a substance capable of operating an aberrant gene product comprising expressing in a cell the gene encoding the aberrant gene product, separating the aberrant gene product, providing a substance as the aberrant gene product and determining operation
10 activity of said substance as said gene product.

25. The method according to claim 24, wherein the aberrant gene product is an aberrant receptor.

26. The method according to claim 24 wherein the substance is a substance that normally operates said product.

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